OBSTETRIX MEDICAL GROUP

Screening or Testing for Fetal Chromosome Abnormalities – LOW RISK Patients

- ☐ Chromosome abnormalities are disorders where the fetus has too much or too little DNA.
- ☐ Depending on exactly which DNA or how much DNA is involved, chromosome abnormalities can range from very mild, barely noticeable problems to extremely severe, with multiple birth defects and no chance of survival.
- ☐ Based on your age and other known factors, you are at LOW RISK of having a baby with a chromosome abnormality, meaning less than 1 in 200 chance (less than ½ of one percent).
- ☐ There are many tests designed to detect fetal chromosome abnormalities. You can choose to have one of these tests or no testing at all. The purpose of this counseling checklist is to help you decide whether to have a test and if so, which test to have.
- ☐ DIAGNOSTIC TESTS will detect *all types* of chromosome abnormalities. These tests will give definite information such as "the chromosomes are normal" or "an abnormality is present". These tests involve actually counting the chromosomes in fetal cells. To obtain fetal cells, it is necessary to obtain a sample of amniotic fluid or placenta using a needle or catheter. There is a small chance that the procedure of obtaining a sample will cause a miscarriage.
- ☐ SCREENING TESTS will detect *some types* of chromosome abnormalities. These tests give information on *risk levels*, such as "the risk for Trisomy 21 is low" or "the risk is high". (Trisomy 21 is the most common chromosome abnormality, also called Down syndrome.) These tests are based on a sample of the mother's blood, so there is no risk of causing a miscarriage.
- ☐ The tests vary in detection rates and rates of women who get a "positive" (abnormal) result.

	Detection Rate for Trisomy 21	Detection Rate for All Chr Abn	Screen Positive Rate	OK for Twins?
Screening Tests				
California 1st trimester screen	80%	69%	5%	Y
California Integrated screen	93%	82%	5%	Y
Cell-free DNA screen	99%	72%	1-9%	N
Diagnostic Tests				
Chorionic villus sampling	>99%	>99%	1%	N
Amniocentesis	>99%	>99%	0.2%	Y

- ☐ Any of the screening tests can have FALSE NEGATIVE RESULTS, meaning that the test result can be "normal" or "low risk" *even if the fetus has a chromosome abnormality*.
- ☐ Any of the screening tests can have FALSE POSITIVE RESULTS, meaning that the test result can indicate a "high risk" of an abnormality even if the fetus has normal chromosomes. For the screening tests, *more than half of the positive (abnormal) results are false positives* in low-risk women, so abnormal results must be confirmed by a diagnostic test.
- ☐ All health insurance plans in California will pay for California Prenatal Screening Program tests, regardless of your risk status (high risk or low risk).
- ☐ For LOW RISK women, some insurance plans will not pay for cell-free DNA screening or for the diagnostic tests. You should discuss your coverage with your insurer if you want one of these tests. If your insurance does not pay, you may be required to pay out-of-pocket for these tests.

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